



SPRED1 gene

sprouty related EVH1 domain containing 1

Normal Function

The *SPRED1* gene provides instructions for making the Spred-1 protein, which helps control (regulate) the Ras/MAPK signaling pathway. The Ras/MAPK pathway is involved in the growth and division of cells (proliferation), the process by which cells mature to carry out specific functions (differentiation), cell movement, and the self-destruction of cells (apoptosis).

The Spred-1 protein attaches (binds) to a protein called Raf, which is part of the Ras/MAPK pathway. The binding of the Spred-1 protein blocks the activation of Raf, stopping the signaling through the remainder of the Ras/MAPK pathway.

Health Conditions Related to Genetic Changes

Legius syndrome

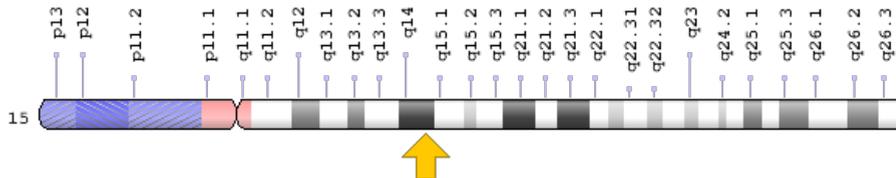
Mutations in the *SPRED1* gene cause Legius syndrome, a condition characterized by multiple café-au-lait spots, which are flat patches on the skin that are darker than the surrounding area.

Several mutations have been identified throughout the *SPRED1* gene. These mutations lead to a nonfunctional Spred-1 protein, usually because the mutated gene provides instructions for a shortened protein. The nonfunctional Spred-1 protein is unable to bind and block the activation of the Raf protein, which means the Ras/MAPK pathway is continuously active. It is unclear how mutations in the *SPRED1* gene cause the signs and symptoms of Legius syndrome.

Chromosomal Location

Cytogenetic Location: 15q14, which is the long (q) arm of chromosome 15 at position 14

Molecular Location: base pairs 38,252,087 to 38,357,249 on chromosome 15 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- EVH1/Sprouty domain containing protein
- FLJ33903
- hSpred1
- NFLS
- PPP1R147
- SPRE1_HUMAN
- spred-1
- sprouty-related, EVH1 domain containing 1
- sprouty related, EVH1 domain containing 1
- sprouty-related, EVH1 domain-containing protein 1
- suppressor of Ras/MAPK activation

Additional Information & Resources

Educational Resources

- University of Utah ARUP Laboratories
http://www.arup.utah.edu/database/SPRED1/SPRED1_welcome.php

GeneReviews

- Legius Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK47312>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SPRED1%5BTIAB%5D%29+OR+%28spred-1%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- SPROUTY-RELATED EVH1 DOMAIN-CONTAINING PROTEIN 1
<http://omim.org/entry/609291>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SPRED1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SPRED1%5Bgene%5D>
- HGNC Gene Family: Protein phosphatase 1 regulatory subunits
<http://www.genenames.org/cgi-bin/genefamilies/set/694>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=20249
- Information Hyperlinked Over Proteins
<http://www.ihop-net.org/UniPub/iHOP/to?focus=IHOP&id=107962>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/161742>
- UniProt
<http://www.uniprot.org/uniprot/Q7Z699>

Sources for This Summary

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